| **Patient Name** | **Patient ID** | **Age** | **Gender** | **Order #** |
|---|---|---|---|---|
| SAMPLEREPORT,SDEL | SA00056048 | 49 | F | SA00056048 |
| **Ordering Phys** CLIENT,CLIENT | | | | **DOB** 08/09/1963 |
| **Client Order #** SA00056048 | **Account Information** | | | **Report Notes** |
| **Collected** 03/21/2013 13:50 | C7028846-DLMP Rochester<br>3050 Superior Drive<br>Rochester, MN 55901 | | | |
| **Printed** 04/16/2013 15:12 | | | | |

| Test | Flag | Results | Unit | Reference Value | Perform Site* |
|---|---|---|---|---|---|
| **Single-gene Large Del/Dup** | | REPORTED 03/26/2013 14:07 | | | |
| Reason for Referral | | | | | MCR |
| Family history of Li-Fraumeni syndrome. Test for the presence of a familial large deletion/duplication in the TP53 gene. | | | | | |
| Result | | | | | MCR |
| The following deletion was detected:<br>Exon(s): 1-11<br>Classification: DELETERIOUS | | | | | |
| Interpretation | | | | | MCR |
| The deletion of exon(s) 1-11 is a known deleterious mutation.<br><br>This result is consistent with a diagnosis of Li-Fraumeni syndrome for this individual. Appropriate screening procedures and/or prophylactic measures should be considered. For information regarding the full tumor spectrum and other associated phenotypes, see GeneReviews for this specific gene (http://www.ncbi.nlm.nih.gov/sites/GeneTests/).<br><br>Since a mutation has been identified, testing of at risk family members is possible.<br><br>A genetic consultation may be of benefit. | | | | | |
| Caution | | | | | MCR |
| CLINICAL CORRELATIONS<br>Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.<br><br>TECHNICAL LIMITATIONS<br>Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered.<br><br>Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.<br><br>EVALUATION TOOLS<br>Multiple in-silico evaluation tools were used to assist in | | | | | |

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|---|---|---|---|---|---|
| the interpretation of these results. These tools are updated regularly; therefore changes to these algorithms may result in different predictions for a given alteration. Additionally, the predictability of these tools for the determination of pathogenicity is currently unvalidated. | | | | | |
| RECLASSIFICATION OF VARIANTS - POLICY<br>All detected alterations are evaluated according to ACMG recommendations (Genet Med 2008:10(4):294-300). Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance. At this time, it is not standard practice for the laboratory to systematically review LIKELY DELETERIOUS alterations or VARIANTS OF UNCERTAIN SIGNIFICANCE that are detected and reported. The laboratory encourages health care providers to contact the laboratory at any time to learn how the status of a particular variant may have changed over time. | | | | | |
| TEST CLASSIFICATION<br>Laboratory developed test. | | | | | |
| Method | | | | | MCR |
| Array comparative genomic hybridization (aCGH) was used to test for the presence of large deletions and/or duplications in the TP53 gene. Mutation nomenclature is based on GenBank accession number NM 000546.4. | | | | | |
| Specimen | | Blood | | | MCR |
| Reviewed By | | | | | MCR |
| Matthew John Ferber PhD | | | | | |
| Release Date | | 26 Mar 2013 14:05 | | | MCR |

* Performing Site:

| | | |
|---|---|---|
| MCR | Mayo Clinic Laboratories - Rochester Main Campus<br>200 First St SW Rochester, MN 55905 | Lab Director: |